

## CLAIMS

What is claimed is:

1. An isolated nucleic acid molecule selected from the group consisting of:
  - (a) a nucleic acid molecule comprising a nucleotide sequence which encodes a polypeptide comprising the amino acid sequence set forth in SEQ ID NOs: 2 or 13; and
  - (b) a nucleic acid molecule comprising a nucleotide sequence which encodes a naturally occurring allelic variant of a polypeptide comprising the amino acid sequence set forth in SEQ ID NOs: 2 or 13, wherein said allelic variant binds to an antibody that selectively binds to the polypeptide of SEQ ID NOs: 2 or 13, and is not the polypeptide within the amino acid sequence of SEQ ID NO: 4.
2. The isolated nucleic acid molecule of claim 1 comprising the nucleotide sequence set forth in SEQ ID NOs: 1, 3 or 12.
3. An isolated nucleic acid molecule comprising a nucleotide sequence which is complementary to the nucleotide sequence of the nucleic acid molecule of claim 1.
4. An isolated nucleic acid molecule comprising the nucleic acid molecule of claim 1 and a nucleotide sequence encoding a heterologous polypeptide.
5. A vector comprising the nucleic acid molecule of claim 1.
6. The vector of claim 6, which is an expression vector.
7. A host cell transfected with the expression vector of claim 6.
8. A method of producing a polypeptide comprising culturing the host cell of claim 7 in an appropriate culture medium to thereby produce the polypeptide.

9. An isolated polypeptide selected from the group consisting of:  
(a) a polypeptide comprising the amino acid sequence of SEQ ID NOs: 2 or 13; and

(b) a polypeptide comprising a naturally occurring allelic variant of a polypeptide comprising the amino acid sequence set forth in SEQ ID NOs: 2 or 13, wherein said allelic variant binds to an antibody that selectively binds to the polypeptide of SEQ ID NOs: 2 or 13, and is not the polypeptide with the amino acid sequence of SEQ ID NO: 4.

10. The polypeptide of claim 9, further comprising at least one heterologous amino acid sequence at the amino- and/or the carboxyl-terminus of said polypeptide.

11. An antibody which selectively binds to a polypeptide of claim 9.

12. A method for detecting the presence of a polypeptide of claim 9 in a biological sample comprising:

(a) contacting said sample with a compound which selectively binds to the polypeptide; and

(b) detecting the presence of a complex between said compound and said polypeptide.

13. The method of claim 12, wherein the compound which binds to the polypeptide is an antibody.

14. A kit comprising a compound which selectively binds to a polypeptide of claim 9 and instructions for use.

15. A method for detecting the presence of a nucleic acid molecule of claim 1 in a biological sample comprising:

- (a) contacting said sample with a nucleic acid probe or primer which selectively hybridizes to the nucleic acid molecule; and
- (b) detecting the presence of a complex of said nucleic acid molecule and said probe or primer.

16. The method of claim 15, wherein the sample comprises mRNA molecules and is contacted with a nucleic acid probe.

17. A kit comprising a compound which selectively hybridizes to a nucleic acid molecule of claim 1 and instructions for use.

18. A method for identifying a compound which binds to a polypeptide of claim 9 comprising:

- (a) contacting said polypeptide, or a cell expressing said polypeptide with a test compound; and
- (b) determining whether the polypeptide binds to the test compound.

19. The method of claim 18, wherein the binding of the test compound to the polypeptide is detected by a method selected from the group consisting of:

- (a) direct detection of binding;
- (b) detection of binding using a competition binding assay; and
- (c) detection of binding using an assay for ABCG4 transporter activity.

20. A method for modulating the activity of a polypeptide of claim 9 comprising contacting said polypeptide or a cell expressing said polypeptide with a compound which binds to the polypeptide in a sufficient concentration to modulate the activity of the polypeptide.

21. A method for identifying a compound which modulates the activity of a polypeptide of claim 9 comprising:

- (a) contacting a polypeptide of claim 9 with a test compound; and
- (b) determining the effect of the test compound on the activity of the polypeptide to thereby identify a compound which modulates the activity of the polypeptide.

22. A method for detecting an allelic variant of the nucleic acid of SEQ ID NOs:1 or 12 or an orthologue thereof in a biological sample, comprising:

- (a) obtaining from the sample a polynucleotide that hybridizes to the nucleic acid of SEQ ID NO:1 or the orthologue thereof; and
- (b) determining whether said polynucleotide is identical to a portion, or the full length sequence, of SEQ ID NOs: 1 or 12, or the orthologue thereof.

23. A composition comprising a pharmaceutically effective amount of the nucleic acid molecule of SEQ ID NOs:1 or 12 and a pharmaceutically acceptable carrier.

24. A composition comprising a pharmaceutically effective amount of an antisense oligonucleotide capable of specifically hybridizing to the nucleic acid sequence of SEQ ID NOs: 1 or 12 and a pharmaceutically acceptable carrier.

25. A transgenic knockout mouse whose genome comprises a homozygous disruption in its endogenous ABCG4 gene, wherein said homozygous disruption prevents the expression of a functional ABCG4 protein, and wherein said homozygous disruption results in said transgenic knockout mouse being sterile.